What is Facioscapulohumeral Muscular Dystrophy?

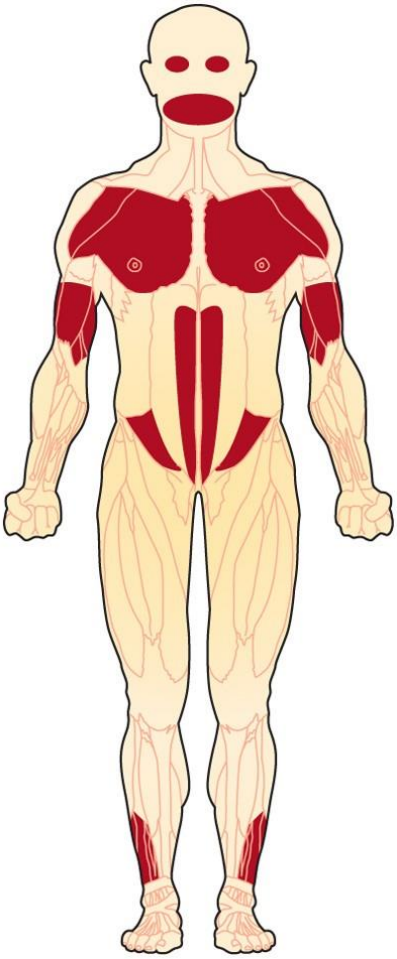
Facioscapulohumeral dystrophy (FSH), is the third most common of nine muscular dystrophy disorders, affecting approximately 1 in 20,000 Caucasians. The term Facioscapulohumeral uses three Latin words to describe the characteristic features of the disorder. Facio means face, scapula means shoulder blade, and humorous is Latin for the upper arm. Muscular dystrophy refers to muscle weakness and wasting. Thus, in FSH the muscles typically affected are those of the face, shoulder blade and upper arms.

Features of FSH

Symptoms of FSH can be classified into two groups: adult-onset, which is usually mild; congenital-onset, which is typically more severe and often present from birth. As well as variability of onset and severity, there is also variability in which areas of the body are affected. The classical symptoms of FSH involve the muscles of the face, shoulder-blade, and upper arms. Some people, however, have no symptoms in the face muscles, with the lower limbs affected instead. This is known as 'Atypical FSH'.

The following features may be displayed:

- Facial weakness, causing difficulty in pronouncing words, and using facial muscles – such as in whistling, smiling or closing one's eyes.
- Weakness in the shoulder blades, preventing movements such as throwing objects and raising one's arms. Lack of strength around the shoulder blades may allow it to wing out.
- Weak lower leg muscles, causing difficulties in walking up hills, or on uneven surfaces. Foot drop may occur when the muscles weaken to such an extent the front part of the foot cannot be lifted up during walking.
- Excessive spinal curve (lordosis), due to abdominal muscle weakness.
- Weakness in the muscles surrounding the hip and those of the upper leg, causing problems when rising from a chair, climbing stairs, or running.
- The development of contractures, as scar tissue replaces normal elastic tissue. This prevents normal movement in the joint, usually the ankles.
- Eye problems are infrequent, but more severe forms of FSH are associated with Coat's disease of the retina.
- Hearing difficulties are common; complete hearing loss may occur in severe FSH.
- Epilepsy in more severe FSH cases.
- Cardiac and respiratory problems are rare in FSH but do occur in some patients.



• Inflammation of the muscles. This can be a source of pain in FSH, as can the altered joint position resulting from muscle weakness.
• Intellectual and cognitive (understanding) difficulties are very uncommon in adults with FSH. There are many FSH people, including those in whom symptoms began in childhood, in intellectually demanding occupations.
• Learning difficulties will on occasion occur, but it is important that real cases are not confused with apparent non-responsiveness. Very bright FSH children may appear intellectually challenged due to a combination of deafness and facial immobility.

FSH tends to progress slowly, and there may be long periods where relatively little change in symptoms occur. It may take thirty years for serious problems to develop, if at all. Approximately 10-20% of FSH patients require a wheelchair, and this may only be occasional. Life expectancy is that of the general population.

This diagram shows the muscle groups affected by FSH. Sourced from: http://mda.org/disease/fshmuscular-dystrophy/overview

What causes FSH?

Unlike other muscular dystrophies such as Duchenne, the mutations causing FSH is not in a gene, but causes a deletion in a part of DNA called a repeat sequence. The size of the deletion is related to the severity of FSH, with large deletions causing congenital onset

(babies born with FSH) and severe symptoms. Small deletions on the other hand, result in later onset (between 15-23 years), and mild symptoms.

FSH is an autosomal dominant disorder in as many as 70 - 90% of cases, meaning only one copy of the defect is required for the FSH to develop. This is in contrast to an autosomal recessive disorder, where two copies of the defect are required for the disease to develop.

The remaining 10-30% of defects arise spontaneously, with the deletion occurring by chance in the egg or sperm at conception. With a spontaneous mutation, the affected person will be the first in his family to have FSH.

There is a 50% chance the child of a FSH parent will inherit the mutation and therefore develop the condition. Females are affected just as frequently as males, although symptoms in men are generally more severe and occur at a younger age than in women.

For further information on genetics and how disorders are inherited, please refer to the Muscular Dystrophy Association Genetics Fact sheet.

Diagnosis of FSH

There are often difficulties in diagnosing FSH, as signs and symptoms of the disease vary. However, once FSH is suspected, diagnostic tests will be offered to establish a definite diagnosis. These may include:

• DNA Testing
There is now a reliable DNA test for FSH, which is approximately 98% accurate as a presumptive diagnosis. Laboratory technicians are able to extract DNA from a small amount of blood and detect the DNA deletion responsible for the disorder. DNA testing can be done during pregnancy to determine if the fetus has inherited the deletion for FSH, but the deletion size cannot be used to predict accurately the severity of the condition. The testing can raise ethical issues for parents faced with the option to terminate a pregnancy.

• CK Testing
A blood test can assess the presence of an enzyme creatine kinase, also known as creatine phosphokinase. This enzyme is usually restricted to muscles cells, but when muscles are damaged as in FSH, the enzyme leaks out and into the blood serum. The CK test will show elevated amounts in the blood but is inconclusive as elevated CK is also a feature of other forms of muscular dystrophies, such as Duchenne.

• Electromyogram (EMG)
An EMG measures the electrical activity of muscles and measures the muscle's response to stimulation of its nerve supply. The results may be nonspecific, or show both nerve and muscle involvement, which is typical of FSH.

• Muscle Biopsy
While under local anaesthetic, a small amount of muscle tissue is taken with a needle, usually from the thigh. Using special staining techniques in the laboratory, the muscle tissue

is examined microscopically. This can give a lot of information on the condition of the muscle, and can help to rule out other diagnoses, or confirm the FSH diagnosis.

Other tests include nerve conduction velocity, hearing tests and tests of cardiac function.

Soon after a diagnosis of FSH in the family, it is essential that genetic counselling is arranged, for one or both of two issues. The first is the probability of Mum or Dad having the disorder and the second is whether testing for FSH in pregnancy can be offered and with what degree of accuracy. Genetic counselling provides information about possible diagnostic tests, including prenatal testing. Genetic services in NZ are available and a referral can be made by the MDA.

Management of FSH

As yet, there is no known cure that can halt or reverse the symptoms and progressive muscle weakness associated with FSH. It is possible, however, to control complications by adhering to a management programme specially designed by a team of medical professionals. This team may include occupational therapists, physicians, orthopaedic surgeons, physical therapists, orthotists, dietitians, nurses and psychologists. Many other people are there to give advice and help in any way possible, such as social workers, teachers, religious advisers, staff from the MDA, parents, and other persons with FSH.

• Exercise
Moderate exercise, especially swimming, is generally considered to be beneficial in FSH, maintaining both muscle strength and flexibility, without undue strain. Approximately 10-20% of those with FSH will at some point be confined to a wheelchair, and swimming is a great source of active exercise for these people, as they are able to use the water for support. Passive exercise, or assisted stretching, should be established as early as possible. Physiotherapists will be able to assist in the development of an exercise programme to delay the shortening of muscles (contractures), which causes limitations in the range of motion of joints. These exercises should be undertaken on a daily basis and require assistance from parents and/ or caregivers.

• Supportive Equipment
Braces and splints are likely to be required to help compensate for weakened muscles. These are often worn at night to help maintain joints in a normal position. Other types of supportive equipment will be available as the need arises, and usually caters to individual need. Advice concerning these will be offered by the physiotherapist, occupational therapist, or by the MDA.

• Medical Treatment
Drugs such as albuterol, clenbuterol, and oxandrolone are being studied for their muscle building effects. These treatments seem to be more effective in the early stages of FSH, improving some measures of strength. Anti-inflammatory drugs may be prescribed to reduce associated inflammation.

• Nutrition

Excessive weight gain leading to obesity can occur due to reduced physical activity produced by muscle weakness. Any excess weight will contribute to tiredness and weakness; hence it is important to maintain a good balanced diet incorporating plenty of fresh fruit and vegetables. Friends and family can assist by restricting foods containing high levels of sugar and fat.

• Surgery
Stabilizing the shoulder blades is one of the more common surgical procedures undertaken by FSH patients. The winged scapulae are fixed to the ribs, so they don't move around. Although the surgery may decrease the arm's range of motion (since the shoulder blade can no longer rotate normally), the ability of the arm to function may be better, as the arm's leverage point is now stable. Tendons can be surgically severed to relieve contractures. This operation is most often performed at the ankle joint but will also benefit those that have already developed severe contractures in the knees and hips. Surgery on the eyelids may be beneficial where there is incomplete closure of the lids. Incomplete closure of the lids may cause inflammation of the cornea (keratitis), so it is important not to ignore the early signs of waking up with dry and irritated eyes. Surgery may produce significant benefits, although these must be balanced against potential complications. Postoperative immobilisation can cause further muscle wasting, and extensive physiotherapy will be required after some surgeries.

Support for people with Facioscapulohumeral Dystrophy

Support is available from the MDA who can offer specialist assessment, information, support, advocacy and referrals to other providers. There is also a nationwide Support Network for those interested in meeting with others.

• Education
In New Zealand, every child has the right of equal access to all aspects of education. This means that all children with a neuromuscular condition have the right to attend a mainstream school. Many schools have special units attached which can provide any extra help needed, including an individualized education plan for appropriate assistance with physical and mental needs.

It is important that FSH children are not overprotected or patronized – they should be mentally stimulated and creative skills encouraged.

• Employment
Seeking and maintaining paid employment can be challenging for people with FSH, especially as their condition progresses. Despite these challenges many people in New Zealand with neuromuscular conditions carve out a career and work productively and successfully for a number of years. Research has shown that a paid occupation is achievable for others with the correct supports and environmental conditions (flexibility, adaptations, employer recognition, peer support).

When choosing a career, if possible choose something that you are passionate about and that meets your physical needs now and into the future as your condition progresses.

Consider the workload; repetitive tasks, physicality of the job, or how much speaking is required if you struggle with slurred speech. Ask about opportunities for job shadowing to get a sense of daily tasks and expectations. Consider when are you more alert and more fatigued? Is there flexibility to work from home on certain days or to be flexible with work schedules so you can incorporate rests if needed? Will the job accommodate flexibility to meet these needs so you can be more productive in your role?

Volunteer work is an opportunity to build up skills and experience. It creates the same feelings of self-worth, sense of identity and purpose as a paid job.

The New Zealand government recognises the value people with a disability can bring to a workforce and the under representation of this community in the labour market. They have set up a number of employment related services and supports for people with a disability, including training and apprenticeships. The list of all government-funded or supported services are available on the website Employment New Zealand.

Diversity Works New Zealand (formally the EEO Trust) is the national body for workplace diversity and inclusion. They can be contacted on 0800 348 377 or by visiting their website diversityworksnz.org.nz/

Remember, it is illegal for employers to discriminate against people because of ethnicity, sexual orientation, gender, marital status, religious belief, or disability. Equal rights are demanded by the Human Rights Act, 1993, and the Equal Pay Act, 1972. You can seek information about your rights on Health and Disability Commissioner website or Human Right Commission website.

More information: Muscular Dystrophy Association can be contacted for further information, assistance, advice, support and referrals, on 0800 800 337 or by e-mail at info@mda.org.nz. The Muscular Dystrophy Association Website also contains information on services available within NZ, our quarterly magazine, contacts, membership details, news and links to other sites - www.mda.org.nz